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ORIGINAL ARTICLE

- Changing Pattern Of Embryonic Heart Rate With Gestation Age: A Marker Of Maturation Of Vagal Activity
- Study Of Thalassemias And Hemoglobinopathies In Pregnant Females In Rural Areas Of North Western India

REVIEW ARTICLE

- Detecting The Deceptive Gossypiboma Post-Vaginal Hysterectomy Using Multimodality Imaging - A Diagnostic Dilemma!

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- Clinico-radiological quiz: Hypoplastic Rib

CPC UPDATE

CASE REPORT

- Prune Belly Syndrome
- A Case Report on Mayer Rokitansky-Kuster-Hauser (MRKH) Syndrome



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The RMJ Policy & Guidelines

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Original Article

Changing pattern of embryonic heart rate with gestation age: A marker of maturation of vagal activity

Mukesh Mittal*, Kapil Gupta**, Nidhi Gupta**, Pankaj K. Nitharwal***

ABSTRACT

Introduction: Early detection of a viable pregnancy can be done by transvaginal ultrasonography. Change in embryonic/fetal heart rate with gestational age is an interplay between increasing demand of fetus for nutrients and oxygen and vagal input to fetal heart.

Material and methods: Transvaginal ultrasonography was done on 109 pregnant women with gestational age ranging from 6 weeks to 10 weeks for gestational age and fetal heart rate. Statistical analysis of the data was done by Microsoft excel.

Observations and results: Mean embryonic/fetal heart rate was increased from 126.45 ± 14.8 BPM from 6 weeks of gestation to 144.58 ± 23.94 BPM at 9 weeks followed by a decline to 123.42 ± 3.42 BPM at 10 weeks.

Conclusion: Decline in fetal heart rate followed by an initial rise in it with increasing gestational age can be a marker of fetal vagal activity.

INTRODUCTION

The cardiovascular system is the first system to function in the embryo.¹ Embryonic development of the fetal heart results from splanchnopleuric mesoderm that form a horseshoe shaped area in the anterior part of the embryonic disk which is a part of the secondary yolk sac wall. In this region two midline endothelial heart tubes form at 18 to 19 days after conception. These endothelial heart tubes fuse to form a single heart tube at about 22 days after conception; 5 weeks 1 day of pregnancy.¹

The earliest proof of a viable pregnancy is obtained when cardiac activity of the embryo can be observed.² High-frequency vaginal transducers have improved embryonic imaging in early pregnancy and have facilitated the very early detection of cardiac activity.^{3,4} Several investigators^{5,6,7} have reported a gradual increase in embryonic heart rate as detected by ultrasonography starting in the fifth week of pregnancy followed by a decline in it. Studies have shown that the baseline fetal

heart rates gradually decreased from 175 to 180 beats per minute(BPM) in the 9 to10 weeks of gestation to 140 to 145 BPM at 19 weeks of gestation, probably due to maturation of fetal vagal function.^{8,9} Development of neural innervations of fetal heart begins in early embryonic life resulting in maturation of fetal vagal function.¹ The present study was done to determine the fetal heart rate variation in early pregnancy in Indian population as there is paucity of literature of similar study in Indian population

MATERIAL AND METHODS

The present study was a cross sectional descriptive and observational study involving the pregnant women attending the antenatal clinic in SMS Medical College and attached hospital over a period of 11 months. The study was started after obtaining approval from institutional scientific and ethics committee. Written informed consent was taken from all the pregnant females. Transvaginal ultrasonography scan was done on 109 pregnant women with gestational age ranging from 6 weeks to 10 weeks. All pregnant females with single embryo were included in this study. Pregnant women with multiple embryos were excluded from this study. Pregnant women with any medical illness were also excluded from this study. The scan was performed using Philips Affinity 70 G machine and fetal heart rate were recorded using M mode. Gestational age was assessed by measuring CRL in mm. Statistical analysis of embryonic heart rate with gestational age was done.

Statistical analysis

The data was analyzed by Microsoft excel software and mean standard deviation and range of fetal/embryonic heart rate was calculated of each weeks of gestational age of fetus.

RESULTS

In the present study, 109 pregnant women were included in the study. The number of pregnant women with 7 weeks, 8 weeks, 9 weeks and 10 weeks of gestation were 40(36.69%), 24(22.01%), 24(22.01%) and 21(19.26%) (Table 1)

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Table 2 shows the mean standard deviation of embryonic/fetal heart rate as per their gestation age. The fetal heart rate increases steeply from 7 to 8 weeks, flattens from 8 to 9 weeks and declines afterwards.

Table: 1 Distribution of number of women as per gestational age of fetus

Gestation age	Number of women
6 weeks 4 days to 7 weeks to 3 days (7 weeks)	40(36.69%)
7 weeks 4 days to 8 weeks to 3 days (8 weeks)	24(22.01%)
8 weeks 4 days to 9 weeks to 3 days (9 weeks)	24(22.01%)
9 weeks 4 days to 10 weeks 3 days (10 weeks)	21(19.26%)

Table: 2 Distribution of gestation age with Fetal/Embryonic Heart rate

Gestation age	Mean(SD) of Fetal Heart rate	Range of Fetal Heart rate
7 weeks	126.45 (14.8) beats/min	105-160 beats/min
8 weeks	141.5 (20.41) beats/min	107-170 beats/min
9 weeks	144.58 (23.94) beats/min	104-170 beats/min
10 weeks	123.52 (3.42) beats/min	119-132 beats/min

DISCUSSION

Presence of fetal heart pulsations is a major sign of embryonic viability in the pregnancy.⁹ Fetal heart rate measurement by M-Mode on ultrasonography is a routine established practice during antenatal checkup of a pregnant women. Fetal/embryonic heart rate varies with the gestational age of fetus and the reference values of normal range of fetal heart rate for different gestation age is present.⁹ Fetal heart rate is determined by the changing circulatory demands of nutrients and oxygen in the developing fetus and the influence of the vagal activity on the fetal heart.^{1,8} Maturation of the vagal activity due to myelination of vagal nerve act as a brake for increased fetal/embryonic heart rate during progression of gestation age of fetus.¹⁰

The present study shows a steep increase of embryonic/fetal heart rate from 7 to 8 weeks of gestation. From 8 to 9 weeks of gestation the slope of rise in heart rate flattens followed by a decline in heart rate from 9 to 10 weeks of gestation.

Our results are similar to observations of various previous studies. Stefos et al (1998) in their prospective study done on 2045 pregnant women showed a progressive increase in mean embryonic heart rate of 111 ± 14 BPM at 6 weeks of gestation to 157 ± 13 BPM at 9 weeks.⁸ Tannirandorn et al (2000) in their study on Thai women found a progressive increase in mean embryonic heart rate from 124 BPM at 6 weeks to 177 BPM at 9 weeks of gestation. After 9 weeks it declines to 159 BPM at 12 weeks of gestation.¹¹ Hanprasertpong T et al (2006) in their study on 319 women observed an increase in embryonic heart from 6 to 8 weeks of gestation. After a maximum value at 8 weeks, the fetal heart rate declines to the lowest values at 14 weeks of gestation.¹² Tezuka et al

(1991) in their study also observed an increase in mean embryonic heart rate from 97 BPM at 5 weeks to 174 BPM at 9 weeks followed by a decline.¹³ Hertzberg et al (1998) showed an increase in embryonic heart rate from 6 to 8 weeks of gestation followed by a plateau in it at 9 weeks after that it followed a declining trend.⁶ Hethyshi R et al in their study on Indian population also found a steady increase in fetal/embryonic heart rate from 6 to 10 weeks of gestation followed by a decline in it.⁹

From all these studies it is clear that the embryonic/fetal heart rate increases from 6-10 weeks of gestation followed by a decline in it. The minor variations in the results may be due to difference in ethnicity of study population along with different methods of determining cut off values for gestation age. The decline in fetal heart from 9 to 11 weeks of gestation may be due to maturation of fetal cardiac vagal function due to myelination of vagal nerve. Gamble et al (1966) done an electron microscopic study on human fetus observed myelination of peripheral nerves from 12-22 weeks of gestation.¹⁴

CONCLUSION

The change in pattern of fetal heart rate with gestation age can be used as a measure of maturation of fetal vagal function.

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Original Article

Study of thalassemias and hemoglobinopathies in pregnant females in rural areas of north western India

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ABSTRACT

Background: Anaemia during pregnancy is a major contributing factor to childhood anemia. The most common cause of anaemia in pregnancy is nutritional anemia. When anemia of pregnancy becomes unresponsive to therapeutic measures, workup for other causes of anemia needs to be done. Hemoglobinopathies and thalassaemias are the most common single gene disorders in the world. Worldwide, the Hb disorders are responsible for 3.4% mortality in children below 5 years of age. High performance liquid chromatography (HPLC) is a simple and rapid method of detection of different Hb variants. The knowledge of the frequency of common Hb variants encountered in pregnant females in a particular area is important for the formulation of specific diagnostic, preventive and therapeutic strategies. There has been no study documenting the prevalence of hemoglobinopathies in pregnant females of Rajasthan, India.

Aim: The aim of the present study was to determine the common Hb disorders in pregnant females of a tertiary care hospital of Rajasthan, India.

Material & Methods: The study was carried out in India with participants recruited during an antenatal care visit made to participating community health and primary health centres (CHCs and PHCs) in the state of Rajasthan. This study was conducted in the Advanced Hematology & HLA Laboratory, Department of Pathology of a tertiary care center.

Results: A total of 1851 pregnant females had their blood samples collected out of which 500 females' samples were evaluated for HBA, HBA2 & HBF by cyanomethemoglobin method. 51 females out of these 500 tested positive for hemoglobinopathy (10.20%). Out of the 51 hemoglobinopathies, 49 were Beta Thalassemia heterozygous, one HPFH & one HBD Punjab.

Conclusions: The most common Hb abnormality detected in this study was that of β thalassemia

heterozygous (9.8%). Due to the high prevalence of Hb disorders in various regions, the premarital screening must be routinely done for prevention of high-risk marriages.

INTRODUCTION

Anaemia during pregnancy is a major contributing factor to childhood anemia. The most common cause of anaemia in pregnancy is nutritional anemia. When anemia of pregnancy becomes unresponsive to therapeutic measures, workup for other causes of anemia needs to be done.

Hemoglobinopathies and thalassaemias are the most common single gene disorders in the world.^[1] Thalassaemias are characterized by reduced synthesis of one or more globin chains of the hemoglobin (Hb) molecule while hemoglobin variants are a result of production of normal amounts of mutant globin chains.^[1] World Health Organization figures estimate that 7% of the world populations are carriers of a potentially pathological hemoglobin (Hb) gene and every year 60000 thalassaemia babies are born all over the world.^[1]

The prevalence of thalassaemias and hemoglobinopathies varies with geographic locations. It has been estimated that in India, 0.37/1000 fetuses have a Hb disorder.^[2] Worldwide, the Hb disorders are responsible for 3.4% mortality in children below 5 years of age.^[3]

High performance liquid chromatography (HPLC) is a simple and rapid method of detection of different Hb variants. Clinical history and findings of thorough hematologic evaluation, including complete blood count, reticulocyte count and red blood cell morphology are necessary to reach an accurate diagnosis. Family studies are often required to detect a particular Hb variant.^[8]

The knowledge of the frequency of common Hb variants encountered in pregnant females in a particular area is important for the formulation of specific diagnostic, preventive and therapeutic strategies. There has been no study documenting the prevalence of hemoglobinopathies in pregnant females of Rajasthan, India. The aim of the present

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study was to determine the common Hb disorders in pregnant females of a tertiary care hospital of Rajasthan, India.

METHODOLOGY

The study was carried out in India with participants recruited during an antenatal care visit made to participating community health and primary health centres (CHCs and PHCs) in the state of Rajasthan.

Inclusion criteria for screening and consent were as follows: (1) pregnant women between 18 and 40 years of age and capable of giving informed consent; (2) Hb concentration of 7–10.4 g/dL (as per the available point of care laboratory report).

This study was conducted in the Advanced Hematology & HLA Laboratory, Department of Pathology of a tertiary care center. A signed consent form was obtained from all the participants included in the study. A detailed clinical history and family history were obtained from each patient. Blood samples were collected in ethylene diamine tetrachloride acetate vials and analyzed with Sysmex automated cell counter (XN 1000) for complete blood counts.

SAMPLE SIZE:

The sample size was calculated at 95% confidence level assuming 4.61% hemoglobinopathies among pregnant patients as found in reference study³ at the precision of 2% (absolute allowable error). Minimum 433 pregnant women were required as sample size which is enhanced and rounded off to 500 pregnant females expecting 15% attribute.

Sample technique:

11 PHC AND 5 CHC of Jaipur CMHO I and CMHO II were selected randomly and 500 pregnant women were taken according to probability proportionate to size method to make sample representative of Jaipur district.⁸

Material and methods

The tests were performed by high performance liquid chromatography (HPLC) based upon the principles of cation exchange chromatography using the BIORAD VARIANT TM Hb typing system (Variant Beta-Thalassemia Short program) (Biorad laboratories, California, USA). A Hb A2/F calibrator were analyzed at the beginning of each run. The different Hb variants like HbE, S, D and C were identified by using retention time windows, that was defined as the time in minutes from sample injection to the maximum point of the elution peak, which are specified for each of these variants. Normal adult chromatogram shows primarily HbA (97-98%), a small percentage of HbA2 (<3.5%) and traces of fetal Hb (<1%).

The data were analysed using Microsoft excel 2010 version.

RESULTS

A total of 1851 pregnant females had their blood samples collected out of which 500 females' samples were evaluated for HbA, HbA2 & HbF by cyanomethemoglobin

method. 51 females out of these 500 tested positive for hemoglobinopathy (10.20%)

Prevalence of haemoglobinopathies

Haemoglobinopathies	No.	%
Present	51	10.20
Absent	449	89.80
Total	500	100.00

Out of the 51 hemoglobinopathies, 49 were Beta Thalassemia heterozygous, one HPFH & one HBD Punjab Trait. The distribution of different Hb patterns in the study population has been shown in **Table 2**. For each of these groups, the percentage of various Hb detected in HPLC have been shown in **Table 2**. Interpretation of results of HPLC was done on the basis of retention time, percentage of Hb and peak characteristics.

Prevalence of various types of haemoglobinopathies

Types of haemoglobinopathies	No.	%
Hb D Punjab Trait	1	0.20
HPFH	1	0.20
β Thalassemia heterozygous	49	9.80
Normal	449	89.80
Total	500	100.00

Parameters	Hbpathies	N	Mean	SD	Median	Min.	Max.	'p' value*
HB	Present	51	9.51	1.06	9.4	7	12.3	0.285
	Absent	449	9.28	1.49	9.4	4.2	13.1	
MCV	Present	51	65.11	5.46	64.4	55.1	78.4	<0.001
	Absent	449	74.67	6.52	75.1	56.8	96.9	
MCH	Present	51	20.11	1.99	19.8	15.9	24.4	<0.001
	Absent	449	22.83	3.07	23	13.3	32.6	
MCHC	Present	51	30.84	1.00	31.1	28.7	33.4	0.207
	Absent	449	30.51	1.83	30.7	24.3	34.8	
RET He	Present	51	20.92	2.59	20.9	15.2	27.4	<0.001
	Absent	449	23.19	4.72	23.2	12.2	36.5	
Hb By Cynmet hemoglobin	Present	51	8.42	1.10	8.2	6	10.9	0.212
	Absent	449	8.17	1.37	8.2	3.4	12	
HbA	Present	51	82.27	4.60	83.4	54.9	85.2	<0.001
	Absent	449	86.06	1.64	86.3	70.9	88.8	
HbA2	Present	51	5.19	0.89	5.3	0.9	6.6	<0.001
	Absent	449	2.60	0.36	2.6	0.5	6	
HbF	Present	51	1.49	2.40	0.9	0.3	17.3	<0.001
	Absent	449	0.45	0.30	0.4	0	4.1	

*Independent Sample 't' Test

This table compares hematological parameters of pregnant females with & without hemoglobinopathies. Significant differences have been found in MCV, MCH, Reticular Hb, HbA, HbA2 & HbF

DISCUSSION

Haemoglobinopathies pose a major health problem in India especially during pregnancy. It has been estimated that in India, 0.37/1000 fetuses have a Hb disorder.^[1] The data on the prevalence of β-thalassemias and other haemoglobinopathies in pregnant females of India is scarce. Therefore, the present study was undertaken to determine the prevalence of haemoglobinopathies in different caste/ethnic groups using uniform methodology.

Most prevalent hemoglobinopathies include beta thalassemia, HbE, HbD and sickle cell anemia. The

prevalence of beta thalassemia mutations is as high as 17% in some Indian populations. The prevalence of HbD in our country is estimated to be approximately 1.1%. [2]

High performance liquid chromatography (HPLC) is a simple and rapid method of detection of different Hb variants. Clinical history and findings of thorough hematologic evaluation, including complete blood count, reticulocyte count and red blood cell morphology are necessary to reach an accurate diagnosis.

In the present study, the prevalence of Hb disorders in pregnant females of rural India was found to be 10.2%. A study conducted in the southern part of West Bengal has reported a higher prevalence (25%) of thalassemias and hemoglobinopathies. [3] a study conducted in north Indian population has reported an incidence of hemoglobinopathies to be 12.5%. [4] The prevalence rate of Hb disorders was reported to be 7% in Bhopal. [5]

The most common Hb abnormality detected in this study was that of β thalassemia heterozygous (9.8%). Colah et al. reported that nearly 1.5% of the world's population is carriers of β thalassemia. [6] In rural Bengal, the prevalence of β thalassemia trait has been reported to be as high as 10.38%. [7] In central India, the prevalence of β thalassemia trait has been estimated to be 9.59%. [8] However, in Orissa, sickle cell trait was the most common abnormality found. [15] In the present study, sickle cell trait was found in 0.46% cases.

In this study, HbE trait was found in 2.68% cases and E β thalassemia in 1.56% patients. A study conducted in the rural areas of West Bengal reported the prevalence of HbE trait to be 3.86% and that of E β thalassemia, 1.25%. [16] Due to the high prevalence of Hb disorders in various regions of West Bengal premarital screening must be routinely done for prevention of high-risk marriages. [17]

Other variants detected in the present study included HPFH.

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Detecting the Deceptive Gossypiboma post-vaginal hysterectomy using Multimodality imaging - A Diagnostic Dilemma!

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ABSTRACT

Gossypiboma, or retained surgical sponge, is a rare post-surgical complication with very few reported cases due to medico legal purposes. Diagnosis is very challenging due to non-specific symptoms, myriad of clinical presentations and use of surgical sponges without radiological marker in most parts of our country. High index of suspicion in all patients with history of past surgery, use of multimodality imaging and thorough knowledge of radiological findings is of utmost importance to diagnose this condition. Infected collection can also be over diagnosed as gossypiboma by different imaging modalities. Therefore, possibility of infected collection as differential diagnosis should always be kept in mind, while diagnosing a gossypiboma.

We present the case of a 72 year old female with recent history of vaginal hysterectomy presenting with mild abdominal pain and distension. We describe the imaging findings in detail with the step wise use of ultrasound in raising suspicion, and CT and MRI to rule out other differentials to finally come to the probable diagnosis of gossypiboma or less likely infected collection and this was confirmed as infected collection by surgery.

INTRODUCTION

The term gossypiboma refers to a surgical sponge or a laparotomy pad left involuntarily in the body after a surgical procedure and is derived from Latin word “Gossypium” meaning cotton and Swahili word “boma” meaning place of concealment^{1,2}.

This rare surgical complication was first reported by Wilson in 1884³, and since then it has been reported in 1 in 100 - 5,000 surgical interventions and 1 in 1,000 - 1,500 intra-abdominal operations⁴.

Gossypiboma poses a great diagnostic dilemma due to non-specific symptoms, associated complications and thus many cases may be misdiagnosed as a tumor⁵ or abscess⁶ in the abdomen and pelvis⁷.

Most gossypiboma cases are discovered during the first few days after surgery; however, they may remain undetected for many years^{1, 8}. Imaging modalities including plain radiography, ultrasonography (USG) though affordable and easily available may not be sufficient and computed tomography (CT), and magnetic resonance imaging (MRI) may be needed^{1,7,9}. Early and accurate diagnosis of gossypiboma plays a very important role for delivering proper treatment and reducing physical and psychological morbidity and mortality⁷. High index of suspicion and thorough knowledge of imaging appearances are needed to make this possible.

CASE REPORT

A 72 year old lady with history of transvaginal hysterectomy 7 days back presented with progressively increasing lower abdominal pain and abdominal distension. There was no history of significant transvaginal discharge or fever. On clinical examination the lower abdomen was tense and tender with presence of mild guarding. She was referred to the radiodiagnosis department to rule out any post op collection or hematoma.

On ultrasound examination an irregular shaped heterogeneous hypoechoic lesion, measuring 6 x 5 cm, with string posterior acoustic shadowing, was noted lying posterior to the urinary bladder and anterior to the rectum, in close proximity to the vaginal stump. Multiple internal echogenic foci with dirty distal acoustic shadowing, likely air foci were noted in the lesion (Figure 1). No internal vascularity was noted on colour doppler examination (Figure 2). The caudal aspect of the lesion was obscured by the shadow of pubic bone and CT scan was performed for better delineation.

On CT scan, lesion appeared localized, heterogenous with soft tissue density and ill-defined margins. Presence of central air foci was confirmed and few specks of hyper densities (Mean-88 HU) were identified in the lesion, likely blood products (Figure 3). Significant surrounding inflammatory changes showing significant contrast

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enhancement were noted on contrast enhanced images with perirectal and perivesical fat stranding, oedematous wall thickening of adjacent walls of urinary bladder, rectum and small bowel loops (Figure 4). No evidence of any fistulous communication, calcification and metallic density noted. A differential diagnosis of infected post-op hematoma v/s retained gauze piece with surrounding foreign body reaction was made. To rule out the presence of anaerobic bacterial abscess, MRI examination was done.

MRI showed presence of central T1 isointense (Figure 5), T2/STIR hypointense area (Figure 6) with significant blooming on GRE images (Figure 7) suggestive of early sub acute blood products. Surrounding heterogeneous STIR hyper intensity was noted (Figure 8); however, no diffusion restriction was noted on DWI/ADC, ruling out the presence of anaerobic bacterial abscess. Differential diagnosis was gossypiboma or less likely infected collection; patient was operated transabdominally and the findings were confirmed as infected collection by surgery.



Figure 1: Sagittal transabdominal ultrasound image showing an irregular heterogenous hypoechoic lesion (straight arrow) with ill-defined echogenic margins and multiple internal air foci (curved arrow) with dirty distal acoustic shadowing was noted in the pelvis, posterior to urinary bladder, and anterior to rectum (arrowhead). Star- urinary bladder with foley's in situ.



Figure 2: Transverse transabdominal colour doppler image showing no vascularity inside the lesion.

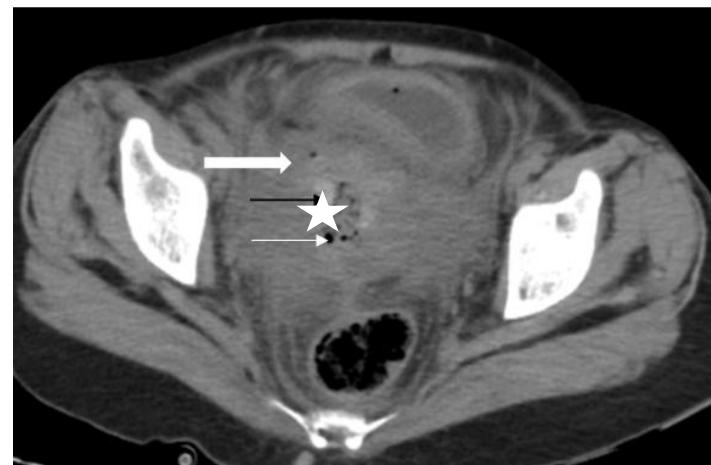


Figure 3: Axial NCCT pelvis image showing localized, heterogenous lesion with soft tissue density (thick white arrow), central air foci (white thin arrow) and few specks of hyperdensities (black arrow) (Mean-88 HU), likely blood products.

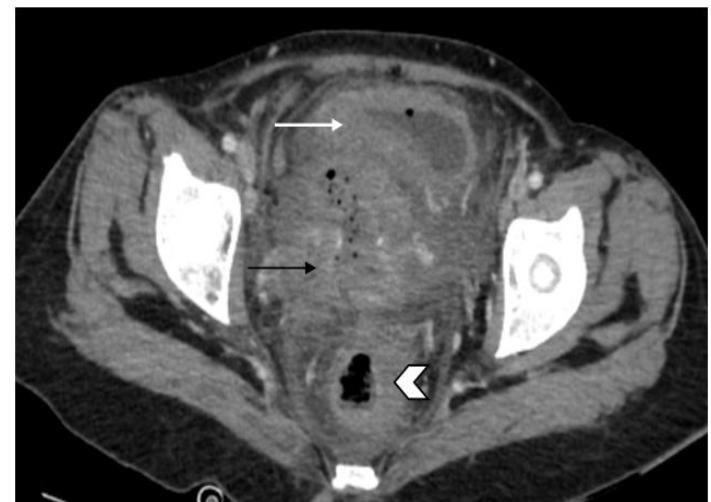


Figure 4: Axial CECT image showing significant perilesional enhancement (black arrow) with thick enhancing posterior urinary bladder wall (white arrow) and anterolateral rectal wall (white arrowhead).

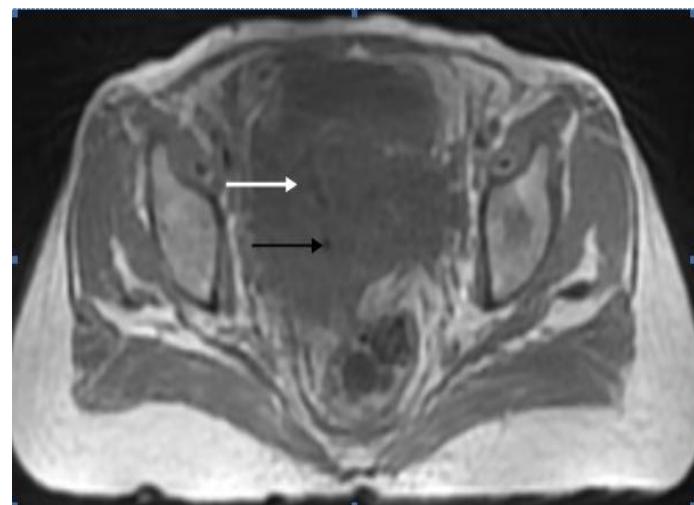


Figure 5: T1 W axial MRI image showing isointense lesion in pelvis with multiple internal hypointense foci.

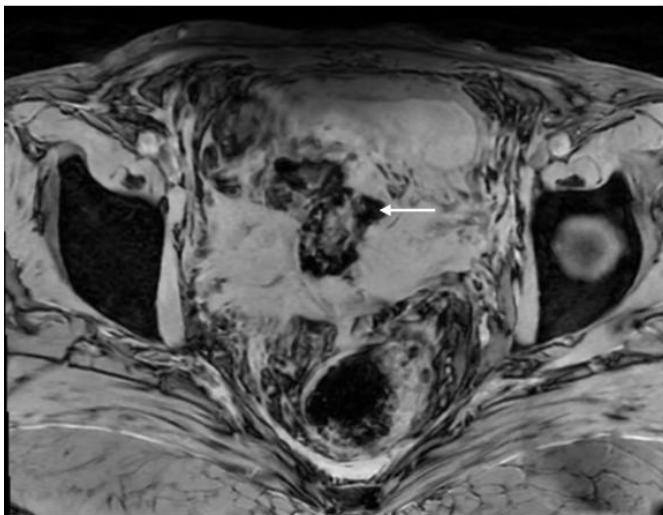


Figure 6: Multiple localized patchy areas of bleeding (white arrow) are noted in the lesion on GRE, suggestive of blood products.

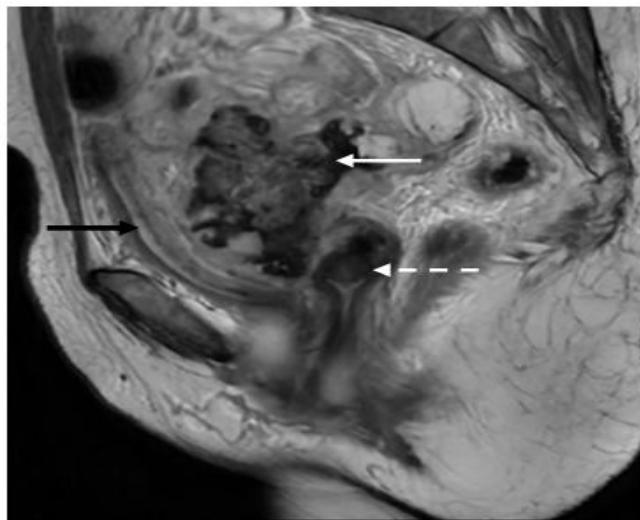


Figure 7: Sagittal T2W MRI image showing T2 hypointensity in the lesion (white solid arrow) lying antero-superior to the vaginal stump (white dashed arrow), and posterior to the bladder (black arrow).

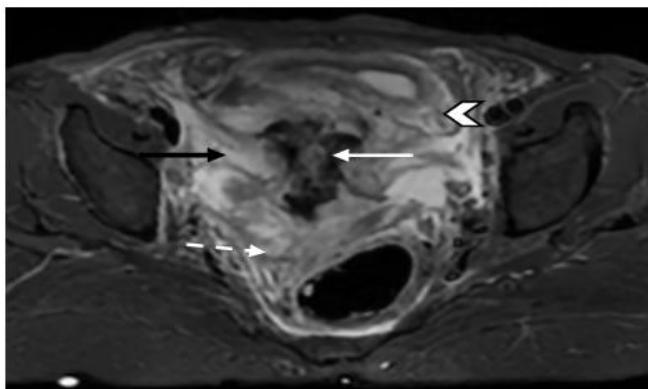


Figure 8: Ill-defined STIR hyperintensity (black arrow) (likely inflammatory tissue) noted surrounding the central heterogeneous hypointensity (white arrow). Also note the increased attenuation of the perirectal (dashed white arrow), perivesical fat and posterior wall of urinary bladder (arrowhead) and anterior wall of rectum.

DISCUSSION

Retained postoperative foreign body is a rare condition, of which surgical sponges are the most common, and is commonly known as gossypiboma or textiloma¹⁰. Abdominal or pelvic cavity is the most frequent location with most cases occurring after gynaecological and upper abdominal surgical procedures¹⁰. Only a few case reports are available till date, describing their imaging characteristics.

Risk factors for development include emergency surgery, unexpected change in the surgical procedure, hurried sponge counts, long operations, unstable patient, obesity etc¹. According to the study by Gawande et al¹¹, retained sponges are 9 times more likely after an emergency operation and 4 times more likely when an unexpected change in the surgical procedure is undertaken¹⁰.

The retained surgical sponge can trigger two types of biological responses: aseptic fibrinous response leading to foreign body granuloma formation or exudative reaction causing abscess formation^{1, 12}.

Gossypibomas may present at any time, from a few weeks to several decades after initial surgery. The symptoms depend upon the location, size of swab, the type of reaction that occurs, time since retention and any associated complications, thus leading to wide variety of presentations⁴. They may present early with pain with or without lump formation or with non-specific symptoms like- nausea, vomiting, abdominal distension, rectal bleeding, altered bowel habit, fever, anorexia, weight loss, mal-absorption syndrome, or a palpable mass^{4, 13}. Others may remain asymptomatic for a long time with only vague symptoms¹.

In chronic cases with intense foreign body reaction, dense adhesions can form around the gossypiboma, and patient may present with complications like sub acute intestinal obstruction¹. Other complications include- abscess formation, peritonitis, fistula formation, non-healing infection of the surgical wound etc⁴. The longer the retention time, the higher is the risk of fistulisation and other complications which highlights the importance of early diagnosis^{4, 14}.

Plain radiography is usually the first investigation done due to wide availability and can help detect gossypiboma containing a radio-opaque marker¹⁰. Radio-opaque threads impregnated into surgical gauzes were first introduced by Cahn in 1929^{4, 15}. The markers may be distorted by folding, twisting or disintegration over time⁴. They are still not used in many parts of our country, and this warrants the use of further imaging to come to a diagnosis⁴.

On ultrasonography, gossypibomas may present as - (i) an echogenic area with intense posterior shadow; (ii) in cases of exudative reactions they are seen as a well-defined cystic mass containing distinct internal hyperechoic wavy, striped focus and (iii) non-specific pattern with a hypoechoic mass^{4, 14}. Acoustic shadowing is

observed in most of the cases likely due to the attenuation of beam by foreign body as well as presence of gas and sometimes calcification^{4,16}.

CT and MRI have superior advantages to ultrasonography in differentiating from tumour, hematoma, or abscess. CT is very useful for identifying radio-opaque marker or gas bubbles present inside the lesion. Calcified reticulate rind sign may be identified in chronic cases, which may be formed by gradual deposition of calcification along the fibre network of the surgical gauze^{7, 17}. The characteristic CT feature is a low-density heterogeneous whorl-like spongiform hypo dense mass containing air bubbles with an external high-density wall showing contrast enhancement^{3,4}.

On MRI, gossypiboma appears as a well-defined mass, showing hypo intensity on T1WI and T2WI^{7,18}. In differentiation of tumor from gossypiboma, DWI plays an important role with lack of diffusion restriction in the gossypiboma¹⁹. As concluded in previous studies, combining patient history and symptoms with imaging features are important to make the accurate diagnosis¹⁰.

Gossypiboma is usually asymptomatic and have nonspecific radiological findings. Therefore, gossypiboma should be included in differential diagnosis of soft tissue masses or localised abdominal pain in patients with history of operation¹.

Treatment protocol is removal of the retained sponge surgically, endoscopically or laparoscopically to prevent severe complications¹⁰.

CONCLUSION

Gossypiboma is a rare postoperative complication which often goes undetected for long duration due to nonspecific symptoms and a myriad of imaging appearances resulting in significant patient morbidity and mortality. It can be avoided by vigilant post-op sponge counts and use of textile materials impregnated with a radio-opaque marker. Gossypiboma without a radiological marker poses a great challenge for diagnosis on radiological imaging as it can simulate a hematoma, granulomatous process, abscess formation, cystic masses, or neoplasm. High index of suspicion in all patients with history of past surgery, use of multimodality imaging and thorough knowledge of radiological findings is of utmost importance to diagnose this condition. Infected collection can also be over diagnosed as gossypiboma by different imaging modalities. Therefore, possibility of infected collection as differential diagnosis should always be kept in mind, while diagnosing a gossypiboma.

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CLINICAL PUZZEL

Clinico-radiological quiz: Hypoplastic Rib

Dr. Ambika Sharma**

ABSTRACT

A thirty-year-old male, nonsmoker, presented with complaints of acute onset moderately severe intensity left sided chest pain after lifting heavy weight. A chest radiograph was taken (Figure 1).



Spot the radiological abnormality?

Answer to Clinico-Radiological Quiz

HYPPOPLASTIC LEFT 4TH RIB.

A thirty-year-old male, nonsmoker, presented with complaints of acute onset moderately severe intensity left sided chest pain after lifting heavy weight. Clinical examination was unremarkable. A chest radiograph was taken (Figure 1) which showed short left fourth rib. A computed tomography (CT) chest was done for further evaluation of same. Hypoplastic left 4th rib (short rib) was confirmed with no other pleuroparenchymal or bony abnormalities. Patient had no history of chest trauma and any surgery. It was an incidental finding and not a cause of his chest pain. He had a muscular cause for his chest pain due to heavy weightlifting which was relieved completely after 3 days of painkiller medications.

A short or hypoplastic rib is characterized by reduced anterior extension toward the sternum, often due to early fusion of the epiphyseal growth plate. This anatomical variation affects approximately 16% of the population,

with a higher prevalence on the right side, though it may also occur bilaterally. Typically, this finding is asymptomatic and isolated, with no clinical significance¹⁻³. In some cases, however, short ribs can be associated with skeletal dysplasias, including thanatophoric dysplasia, achondroplasia, Ellis–van Creveld syndrome (chondroectodermal dysplasia), Jeune syndrome (asphyxiating thoracic dystrophy), and short-rib polydactyly syndromes. Awareness of these associations is important when evaluating patients with known genetic or skeletal disorders¹⁻³.

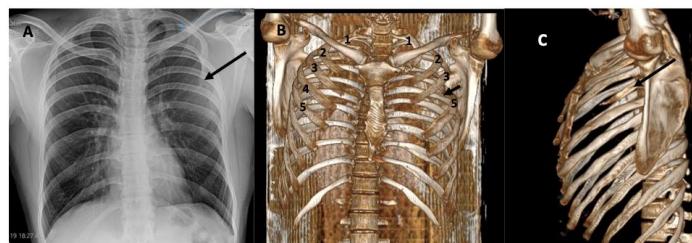


Figure 1. HYPOPLASTIC RIB (Short rib). A- Chest Xray PA View, B- three-dimensional reconstruction image of computed tomography chest; C- three-dimensional reconstruction image of computed tomography chest left lateral view. Showing hypoplasia of the left fourth rib (Black arrow).

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CPC UPDATE

The clinicopathological conference, (CPC) was organized by the MEU on 17 September 2023 aiming to utilize case based study method of teaching medicine. This was presented by the department of medicine. Here are the excerpts from the case.

AN UNUSUAL CASE OF MOVEMENT DISORDER

A 53-year-old Hindu, married female, housewife, graduate, resident of Jaipur presented with chief complaints of Involuntary Perioral Chewing movements, slurring of speech and abnormal finger movements for 10 years which further increased gradually. The movements of right hand were insidious in onset, constant, repetitive and purposeless chewing movements with lip smacking, intermittent opening and closing of jaw. It initially started with episodes of short duration and gradually progressed to remain throughout the day. It disappeared in sleep, as observed by family members and were not altered by eye closure. It remained unaffected by voluntary movements but were affected by emotions/excitement. It was not affected on approaching an object. Slurred speech was insidious in onset, gradually progressive. Patient had Dysarthric type of speech with Normal loudness, Intonation and Low pitch.

There was No history of fall or loss of consciousness in past or weakness of upper and lower limbs and no facial deviation, no history of medications / seizure disorder/ palpitation/weight loss/ headache/vomiting/blurring of vision. Patient was a known case of Type-2 diabetes mellitus; on oral hypoglycemic agents (on glimepiride plus metformin) for last 6 years with good sugar controls. She had Hypertension and was on Anti-hypertensives for last 5 years (Telmisartan and Amlodipine). No history of surgery/hospitalization or any other chronic illness.

On examination, Patient was conscious, co-operative and well oriented to time, place and person, average built and nourished. Pulse rate 84/minute, regular in rhythm, normal volume, vessel wall not palpable, no radio-radial and radio-femoral delay, all peripheral pulses well felt. BP: 130/80 mmHg in right arm in supine position RR: 16/minute, thoraco-abdominal; Temperature: Afebrile by touch. Cranial Nerves were normal on examination.

The examination of the Motor System showed Muscle tone to be increased in right upper limb, rigidity was present (cog wheel type). Power: 5/5 in all four limbs. There was Involuntary movement present in fingers of right Hand-Arrhythmic, repetitive and purposeless, appear at rest, increased on excitement and getting emotional, not suppressible and not altered by eye closure, disappeared during sleep, however, there was no change on approaching an object. Examination of sensory system showed that all intact sensations. Cerebellar functions were intact. Gait showed mild slowness in getting up from sitting, freezing was noted on initiation with reduced step length and arm swing right > left with some distal

choreiform movements in right upper limb. **Cranium and spine and meninges** were within normal limit

She had mildly reduced facial expressions, with a staring look. Blink rate was reduced. Cog-wheel rigidity was more on the right side. Bradykinesia was present right > left. Mild perioral dyskinesia with lip smacking with jaw dyskinesia. Distal choreiform movements were also observed in the right hand. On **Slit lamp examination: No K-F ring was seen**

Clinical diagnosis: **Insidious onset gradually progressive oro-buccal dyskinesia with right distal choreiform movements with dysarthria with bradykinesia and cog-wheel rigidity s/o the extrapyramidal tract involvement with mild cognition and psychiatric involvement. Clinical findings suggest a movement disorder associated with neurodegenerative changes of long duration with gradually progressive course.** Differential diagnosis included Likely a neurodegenerative disorder: Huntington's Disease/ Wilson's Disease/ NBIA/ Neuroacanthocytosis/ Atypical Parkinson's disease.

Routine blood and urine examination were within normal limits. MRI showed discrete and semi confluent old infarct with gliotic areas seen in centrumsemiovale, periventricular white matter and bilateral ganglia showed small vessel ischemic changes suggesting a possibility of pantothenate kinase associated neurodegeneration (PKAN).

DISCUSSION

NBIA (Neurodegeneration with Brain Iron Accumulation) are a heterogeneous group of genetic neurodegenerative disorders characterized by abnormalities in brain iron metabolism and with excess iron accumulation in the **globus pallidus** and to a lesser degree in the **substantia nigra** and sometimes adjacent areas. All of the NBIA disorders feature iron deposition in the globus pallidus but differ in the co-occurrence of other findings

Neurodegeneration with Brain Iron Accumulation: Genetic Types

Gene ¹	NBIA Genetic Type	MOI	% of all NBIA
<i>ATP13A2</i>	Kufor-Rakeb syndrome (OMIM 606693)	AR	Rare ²
<i>C19orf12</i>	Mitochondrial membrane protein-associated neurodegeneration (MPAN) (MPAN)	AR, AD ³	5%-10% ⁴
<i>COASY</i>	COASY protein-associated neurodegeneration ⁵ (CoPAN; OMIM 618266)	AR	Rare
<i>CP</i>	Aceruloplasminemia	AR	Rare
<i>DCAF17</i>	Woodhouse-Sakati syndrome	AR	Rare ⁶
<i>FA2H</i>	Fatty acid hydroxylase-associated neurodegeneration (FAHN)	AR	Rare ⁷
<i>FTL</i>	Neuroferritinopathy	AD	Rare
<i>PANK2</i>	Pantothenate kinase-associated neurodegeneration (PKAN)	AR	30%-35% ⁴
<i>PLA2G6</i>	PLA2G6-associated neurodegeneration (PLAN)	AR	10%-15% ⁴
<i>WDR45</i>	Beta-propeller protein-associated neurodegeneration (BPAN)	XL	40%-45% ⁴

PKAN (autosomal recessive) is a major form of NBIA accounting for approximately 50% of childhood NBIA with a prevalence of 1-3/million. The Clinical features include Extrapyramidal involvement -dystonia rigidity and parkinsons – cranial onset with dysarthria later on involving limbs; Pyramidal tract resulting in upper motor

neuron signs of hypertonicity, hyperreflexia, and spasticity. Pigmentary retinopathy leading to visual impairment. Pattern of stepwise decline, with periods of relative clinical stability combined with episodic neurological deterioration, cognitive decline, and loss of motor skills. It is caused by mutations in the PANK2 gene on chromosome 20. Globally, the c.1561G>A missense mutation is the most common cause of PKAN.

Death is usually secondary to (i) cardiorespiratory complications and complications from malnutrition or status dystonicus.

Very few case reports have been from India, one being Parashari UC, Aga P et al did MR spectroscopy in pantothenate kinase-2 associated neurodegeneration. Panneer Devaraju, Bhuvaneswari Arumugam et all reported an atypical PKAN in one family where 2 siblings, where first sibling symptoms at age of 32 in form of cervical dystonia, blepharospasm and cognitive decline. 2nd sibling had right hand dystonia, dysarthria, dysphagia psychiatric symptoms and cognitive decline. Both had "EYE OF TIGER" in MRI brain.

MANAGEMENT OF PKAN

Currently there are no disease-modifying treatments for any form of neurodegeneration with brain iron accumulation. Treatment options remain supportive and palliative. Multidisciplinary approach with close collaboration between health care professionals is needed. This includes: Neurologic management of extrapyramidal and pyramidal disorders, seizures, and sleep disturbance; neuropsychiatric symptoms;

There are several promising treatments currently under active investigation. These treatments can be grouped under four broad approaches: Iron chelation to treat brain iron deposits; Metabolite supplementation to restore metabolic deficits in CoA pathway Such as Co-A, Cyclic PPA; PANK3 activation; GENE therapy to introduce a functional copy of the PANK2 gene; Gene therapy. Gene therapy would be an attractive option for medically intractable life-limiting NBIA disorders. Deep brain stimulation (DBS) has been undertaken in some patients with NBIA, and mainly in PKAN with intractable dystonia. Overall, it appears that DBS is generally a **safe and well-tolerated procedure** it may be a reasonable option for the palliation of severe pharmacoresistant dystonia.

Take home message

- A. Early diagnosis in movement disorders with neurodegeneration will improve prognosis and aid in genetic testing of proband and counselling for prevention in progeny.
- B. Spectrum of movement disorders associated with brain iron accumulation labelled as NBIA have received a new limelight with development of better techniques of MR imaging.
- C. Understanding of these disorders have also opened new areas for research in understanding iron metabolism in health and diseases of the CNS.

Better understanding of pathology underlying spectrum of NBIA has paved way for newer treatment approaches.

CASE REPORT

Prune Belly Syndrome

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ABSTRACT

Prune Belly syndrome (PBS) is a rare congenital anomaly of uncertain aetiology almost exclusive to males. PBS may be associated with lower urinary tract obstruction (LUTO) which results in bladder distension and abdominal musculature deficiency. Therefore, early detection of the disease and proper treatment before the renal impairment is important which may require intrauterine intervention like vesicocentesis. We report a case of prune belly syndrome diagnosed at 17 weeks of gestation with USG findings of a single live foetus with anhydramnios, over distended urinary bladder covering almost entire abdominal cavity, bilateral hyroureteronephrosis and single umbilical artery.

INTRODUCTION

Prune belly syndrome (PBS) is known as Eagle–Barrett Syndrome or Obrinsky syndrome is a rare syndrome affecting about 1 in 40,000 births and is characterized by a lack of development of abdominal wall muscles giving the appearance of thin wrinkled skin which appears “prune-like”, skeletal anomalies, and renal anomalies such as dilated bladder, megaureters, and bilateral cryptorchidism¹. The exact aetiology of this disorder is not known but some studies have indicated that there is a possibility of genetic inheritance and possible chromosomal association with Edward and down syndrome². More than 95% of affected cases are of male gender.

CASE PRESENTATION

A 27 year second gravida came for first antenatal scan. Previous pregnancy was completely normal. No significant medical or family history. No H/o drug intake. USG findings revealed a single live foetus of about 17 weeks of gestation with anhydramnios, over distended urinary bladder covering almost entire abdominal cavity, bilateral hyroureteronephrosis and single umbilical artery. Other foetal biometry was normal. All bowel loops were pushed cranially towards liver which indicates that lesion is likely of pelvic origin pushing peritoneal organs upwards. Pregnancy was terminated on request of family members and findings of prune belly syndrome were confirmed as foetus was showing thin wrinkled abdominal wall and large ruptured cystic cavity.

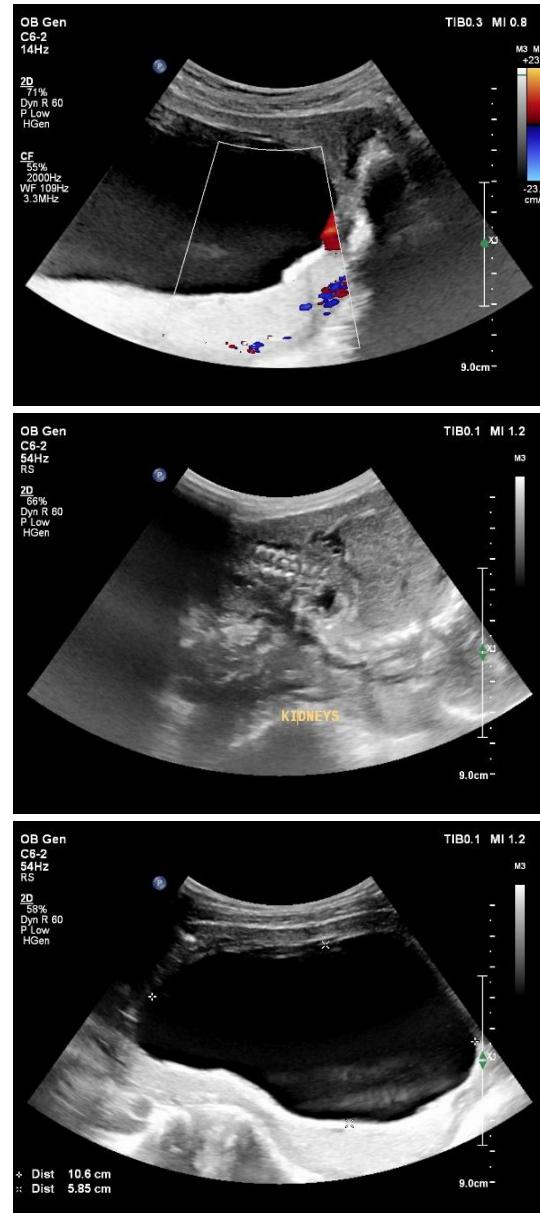


Image 1 A : Color doppler image showing single umbilical artery B : Grey scale image showing well defined cystic lesion in the abdominal cavity. C: Grey scale image showing bilateral hydronephrosis.

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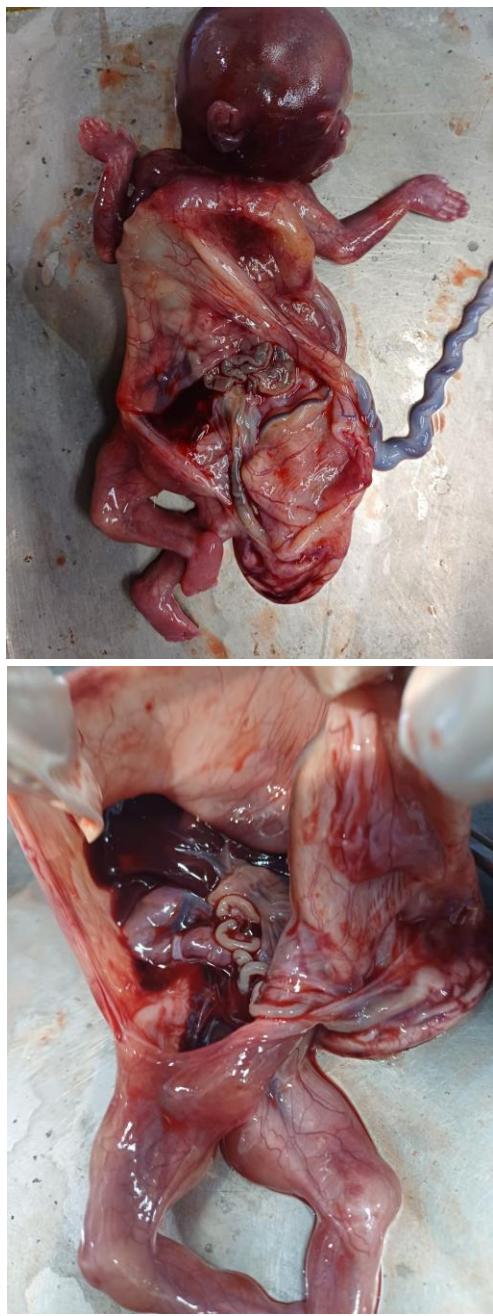


Image 2: Post natal images of the foetus showing thin wrinkled abdominal wall and large ruptured cystic cavity.

DISCUSSION

Prune-belly syndrome or Eagle-Barrett syndrome is a rare congenital anomaly. The etiopathogenesis of PBS is not completely understood. Many theories have been suggested; urinary tract obstruction, genetic deficit, abnormalities of allantois and maldevelopment of the mesoderm. Prune belly syndrome occurs with variable degrees of severity. Those with less severe renal disease may survive infancy, but may have recurrent urinary tract infection or progressive renal insufficiency³. Little or no loss of renal function is seen in some mild cases with a better prognosis. In severe cases, renal dysplasia and oligohydramnios in utero result in pulmonary hypoplasia. These infants may be stillborn or die shortly after birth often due to respiratory distress. Pulmonary,

cardiovascular, gastrointestinal and musculoskeletal abnormalities are also associated with Prune belly syndrome. The clinical manifestations are deficiency of abdominal wall muscles, cryptorchidism in male fetus with genitourinary malformations⁴⁻⁵. Prune belly syndrome is characterized by pulmonary hypoplasia, pneumothorax, oligohydramnios, renal dysplasia, urethral obstruction patent urachus or club feet. Although pulmonary hypoplasia may not be clearly identified in the very early gestational age. Urine production in foetus starts from 8 and 10 weeks of gestation and fetal bladder can be visualized by sonography by 11 weeks of gestation. Therefore the antenatal diagnosis of prune belly syndrome is rarely reported before 11 weeks of gestation²⁻⁴. In our case fetus was diagnosed as prune belly syndrome by prenatal abdominal ultrasound at around 17 weeks of gestational age with very large urinary bladder. As renal function compromise is a factor that determines the prognosis, prenatal intervention is needed to preserve renal function and to provide the aqueous environment for lung maturation⁶. Regular monitoring by sonography of the urinary tract and amniotic fluid volume in utero is required with antenatal management of Prune belly syndrome by fetal vesicoamniotic shunting, termination of pregnancy, or early delivery as long as the neonatal viability is achieved. Vesicoamniotic shunt for urinary obstruction can prevent pulmonary hypoplasia and renal dysplasia or even may improve postnatal quality of life. Vesicocentesis for the treatment of fetal megacystis and prune belly syndrome have been reported to reduce the bladder volume without severe complications. This study suggests the importance of the early diagnosis of this congenital anomaly however in our case, pregnancy was terminated upon request of parents. Furthermore, as prompt intervention in second trimester the vesicocentesis with finer needle can be used as an effective treatment modality to relieve the lower urinary tract obstruction to improve the prognosis.

CONCLUSION

Prune-belly syndrome or Eagle-Barrett syndromes is a rare congenital anomaly. Radiologists must be well versed with this anomaly for antenatal diagnosis as early as possible so that timely decision can be made regarding the course of pregnancy, prenatal intervention to preserve renal function and to provide the aqueous environment for lung maturation and post-delivery management based on the severity of the condition.

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Case Report

A Case Report on Mayer Rokitansky-Kuster-Hauser (Mrkh) Syndrome

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ABSTRACT

Mayer Rokitansky Kuster Hauser syndrome (MRKHS) is a congenital malformation of female genital tract. In MRKHS there is uterine agenesis or hypoplasia of proximal vagina due to interrupted embryonic development of para-mesonephric ducts. We report a case of type 2 MRKHS with left crossed fused renal ectopia. Ultrasonography findings were absent uterus, normal bilateral ovaries, left crossed fused ectopia with left dilated tortuous ureter which was confirmed on MRI. For management and corrective of surgery of patient correct diagnosis is essential.

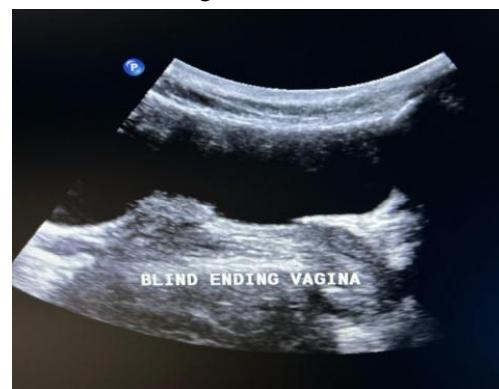
INTRODUCTION

Mayer-Rokitansky-Kuster-Hauser Syndrome (MRKHS) or Mullerian dysgenesis is an infrequent congenital syndrome. Incidence of MRKHS is 1 in 5000 females. It is characterized by uterine agenesis or hypoplasia of proximal vagina due to interrupted embryonic development of para-mesonephric ducts with normal karyotype 46XX and normal secondary sexual characteristics¹. They are of two types: type 1 having only uterovaginal agenesis and type 2 having uterovaginal agenesis with anomalies in fallopian tube, kidney, spine, heart and other organ and amenorrhea with painful sexual intercourse¹. For normal married life counseling of the patient and surgical correction by neo vagina creation for sexual intercourse is the treatment of choice. Here, we report a case of type 2 MRKHS with left crossed fused renal ectopia as per SCARE 2020 criteria². The MRKH syndrome had a significant influence on both fertility and psychological health of women, therefore correct diagnosis with accurate anatomical details is required for counseling of the patient and management by surgical correction.

CASE REPORT

A 21 year old unmarried girl presented to our hospital with the chief complaints of primary amenorrhea. She had normal secondary sexual characteristics. There was no family history of primary amenorrhea. There was no history of antenatal exposure to her mother for any medication. Patient had no past medical or surgical history.

Trans-abdominal ultrasound scanning of patient showed absent uterus, normal bilateral ovaries showing non-dominant follicles, left crossed fused ectopia and dilated and tortuous left ureter. MRI on T2W image showed left dilated tortuous ureter with left cross fused renal ectopia and blind ended vagina with normal bilateral ovaries.



A.



B.



C.

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Figure-1: USG grey scale images showing A - Absent uterus with blind ended vagina B- Dilated tortuous left ureter C - Normal left ovary D - Normal right ovary E - Left cross fused renal ectopia

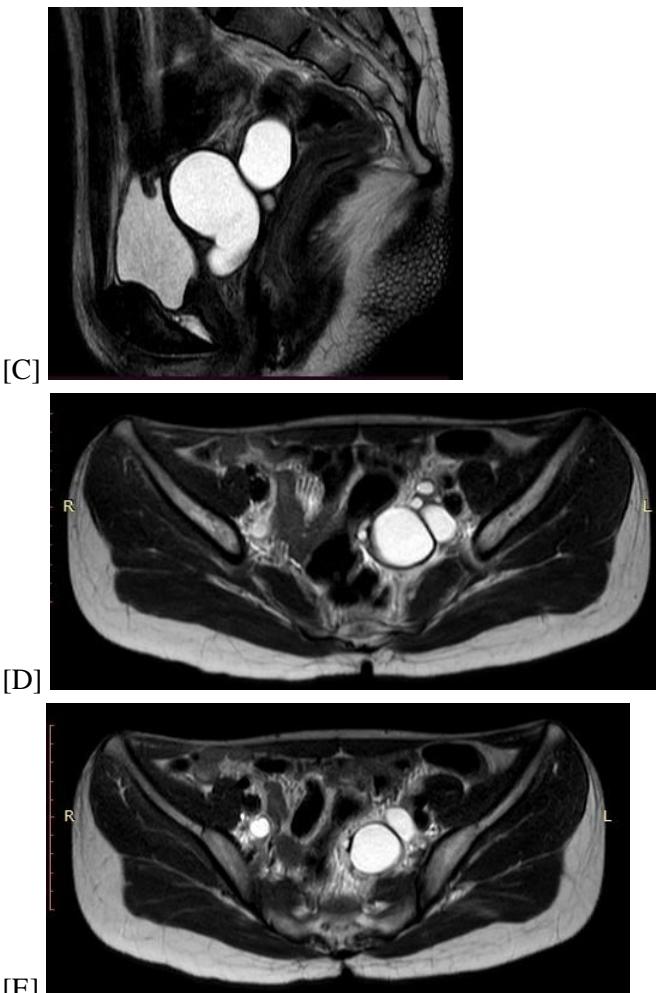


Figure-2: MR T2W images showing A - Left dilated tortuous ureter B - left cross fused renal ectopia C- blind ended vagina D - Normal left ovary E – Normal right ovary

DISCUSSION

Mayer-Rokitansky-Kuster-Hauser syndrome (mullerian agenesis) is a spectrum of congenital anomalies with no known exact cause however mutation in WNT4 gene may be the cause of Mullerian aplasia³. In MRKHS, patient has varying spectrum of utero-vaginal agenesis with normal ovaries and normal secondary sexual characteristics¹. Uterus and proximal vagina may be absent or rudimentary as bilateral and non-cannulated muscular buds. Patient has bilateral normal ovaries and fallopian tubes with normal endocrine and cytogenetic evaluations⁴. This case presented with mullerian agenesis and left crossed fused renal ectopia which is classified as MRKHS type 2 association. MRKHS type 2 may be rarely associated with anomalies of upper urinary tract, skeletal and cardiac defects¹.

Urinary tract malformations are seen in 40% of the cases which mainly includes unilateral renal agenesis, hypoplastic kidneys, horse shoe shaped kidneys and hydronephrosis¹. Skeletal anomalies are seen in 30–40% of the cases¹. It includes scoliosis, isolated vertebral anomalies, ribs malformations, and spina bifida. Involvement of face and limbs is rarely seen¹. Limb



abnormality includes ectrodactyly, duplicated thumb¹. Auditory defects are seen in 25% of MHRC type 2 cases with conductive deafness due to stapedial ankylosis due to middle ear malformations or sensorineural defects of varying severity¹. Cardiac malformations may be tetralogy of fallot, atrial septal defect and conotruncal defects like pulmonary valvular stenosis¹. The diagnosis of MRKH mainly depends on imaging study. Transabdominal ultrasonography is the first line investigation but abdomino-pelvic MRI gives more precise and clear information than the sonography⁵.

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